Newborn Screening Section- History Synopsis

The State of Michigan introduced newborn screening in 1965, when laboratory technology to identify newborns with phenylketonuria (PKU) first became available. In 1977, a test for congenital hypothyroidism (CH) was added, and in 1985, galactosemia screening was initiated. Public Act 14 of 1987 mandated further expansion. This Act added three disorders; biotinidase deficiency, maple syrup urine disease (MSUD), and hemoglobinopathies (sickle cell disease). It also designated the state laboratory as the sole testing site, mandated a fee to fund the program, added comprehensive follow-up, medical management centers, and quality assurance. In July 1993, the laboratory began testing for congenital adrenal hyperplasia (CAH).

Tandem Mass Spectrometry (TMS) technology was implemented in April 2003 when medium chain acyl-CoA dehydrogenase deficiency (MCAD) was added to the screening panel. The existing assays for MSUD, PKU and were switched to TMS at that time. Homocystinuria, citrullinemia, and argininosuccinic aciduria were three additional disorders screened by TMS that were added in October 2004. Information can be found on the disorders screened at this time.

A pilot project was initiated in April 2005 that was authorized by the Newborn Screening Advisory Committee to evaluate the feasibility of expanding the panel of eleven disorders to include twenty-nine additional amino acid, fatty oxidation, and organic acid disorders by TMS. In May 2006 the laboratory report was modified to reflect the new disorders screened.

When an infant screens positive for one of the conditions, the Newborn Screening Laboratory notifies the follow-up office in the Bureau of Epidemiology, Epidemiology Services Division. Lab Scientists also phone/fax one of the state-contracted medical management centers. For a metabolic condition, such as PKU or MCAD, the Children's Hospital of Michigan Metabolic Clinic is notified. If the test is positive for CH or CAH, the staff members would contact the Pediatric Endocrine Center. The Sickle Cell Disease Association of America, Michigan Chapter, Inc. is notified about infants that screen for sickle cell or related diseases. The physician responsible for care of the newborn is also informed. Depending on the test result, rescreening may be required, a serum test may be ordered, or, rarely, immediate referral to a specialist or hospital emergency room is ordered.

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